



THE HUMAN VARIOME PROJECT

Human Variome Project Bulletin No. 12

Nov 07 - Feb 08

1. Activities in the area of “mutation collection”/HVP have had a dramatic boost in many areas over the last four months which is exciting.
2. Congratulations to the GEN2PHEN team led by Tony Brookes for getting EC funding to build a system into which the rest of us can deliver variation. This is exciting news for the field.
3. Sean Tavtigian hosted a meeting at IARC in Lyon in February on unclassified variants (“Pathogenic or not”), which included major experts in the field. He and Marc Greenblatt will edit an issue of *Human Mutation* with eight papers arising.
4. We congratulate Victor McKusick the major originator of our field and strong supporter of the HVP on his award of the Japan Prize.
5. A Human Variome Project planning meeting is to be held on the Costa Brava, Spain May 25-29, 2008 (www.humanvariomeproject.org/HVP2008/). This will coincide with a HGVS Genotype to Phenotype meeting (www.hgvs.org/Barcelona08/) on May 31st, which is a satellite of the ESHG meeting in Barcelona (www.eshg.org/ESHG2008/). Those interested should contact the organizers shown on the websites.
6. It is important that those unable to attend the Costa Brava meeting who have systems, plans and ideas relevant to collection of mutations and their effects, should send an abstract so discussion leaders can include them in the discussion and later publication.
7. To popularize the HVP especially amongst the younger generation, we have started a Facebook page. Please have a look (<http://www.facebook.com/pages/The-Human-Variome-Project/10227370177>) and give us your comments. Thanks to Russ Altman for becoming an early fan!
8. Myles Axton/*Nature Genetics* has continued his welcome support of the HVP and has been consulting widely on the topic of Microattribution and has two recent Editorials in *Nature Genetics* August 07 and January 08. He will be presenting a plan at the planning meeting in Costa Brava.
9. We are fortunate to have Myles Axton visit Melbourne the week of March 10th and have generated a minor workshop (www.humanvariomeproject.org/?p=Melbourne_meeting) and we expect to feed ideas and plans from this meeting for consideration in Costa Brava.
10. Discussion has been occurring between the “LSDB community” regarding conditions of sharing of data with central database/Browsers and a discussion document is near complete.

11. The HUGO Gene Nomenclature Committee recently announced that they have added links to the LSDBs, the listing of which is curated by Connie Talbot, Rania Horaitis and Many Phommarninh (www.hgvs.org/dblist/glsdb.html).
12. "Ultra-fast genomics conference" of interest 8-10 December 2008, Dubai (www.ultrafastgenomics.com).
13. Papers of interest:
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 - Axton, M. Human Variome Microattribution Reviews. *Nat Genet* **40**(1): 1 (2008).
 - Giles, J. Looking behind the numbers. *New Scientist* **197**(2643): 44-5 (2008).
 - Capriotti, E., Arbiza, L., Casadio, R., Dopazo, J., Dopazo, H. and Marti-Renom, M. A. Use of Estimated Evolutionary Strength at the Codon Level Improves the Prediction of Disease-Related Protein Mutations in Humans. *Hum Mutat* **29**(1): 198-204 (2008).
 - Wildeman, M., van Ophuizen, E., den Dunnen, J.T. and Taschner P.E.M. Improving Sequence Variant Descriptions in Mutation Database and Literature Using the Mutalyzer Sequence Variation Nomenclature Checker. *Hum Mutat* **29**(1): 6-13 (2008).
 - Reddington, F., Ajose-Adeogun, A. and Clark, R. The UK National Cancer Research Institute (NCRI) Informatics Initiative: promoting partnership in cancer research. *Hum Mutat* **28**(12): 1151-1155 (2007).

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